COLLABORATIVE 'BRAIN IN A DISH' PROJECT TO SEARCH FOR NEW TREATMENTS FOR SANFILIPPO SYNDROME

The Sanfilippo Children's Foundation has secured $2million of MRFF funding and engaged leading researchers at three partner institutes for a major drug screening project set to fast-track research towards effective treatments for the rare and terminal childhood dementia, Sanfilippo Syndrome.

Researchers from the South Australian Health and Medical Research Institute (SAHMRI), Adelaide's Women's and Children's Hospital and the University of Adelaide – in partnership with the Sanfilippo Children's Foundation – will create patient-specific neuronal cell models that will be used to search for drugs to address Sanfilippo Syndrome.

Associate Professor Kim Hemsley from SAHMRI's Lifelong Health theme says the technology involves taking skin cells from patients, reverse engineering them into induced pluripotent stem cells (iPSCs) and then developing them into neural cells – an individualised representation of the person's brain.

"Each of us reacts differently to a given medication so by using a patient's own cells we can create a targeted, personalised treatment plan," A/Prof Hemsley said.

"The other benefit is we can grow a significant number of these 'brains in dishes', meaning we can fast-track the testing of a range of drugs that have already been given safety approval for human use."

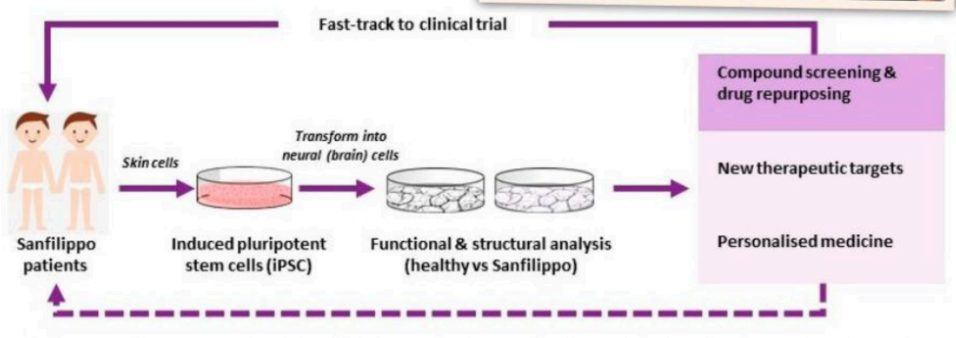
> ***"Testing using a patient's own cells fast-tracks the research because it enables multiple drug combinations to be trialled rapidly and without risks to the children themselves."***

They will test many hundreds of different drugs on the cells, initially this will include drugs that are already used for other conditions and could potentially be repurposed for Sanfilippo, but the models will also be useful for testing new therapies being specifically developed for Sanfilippo.

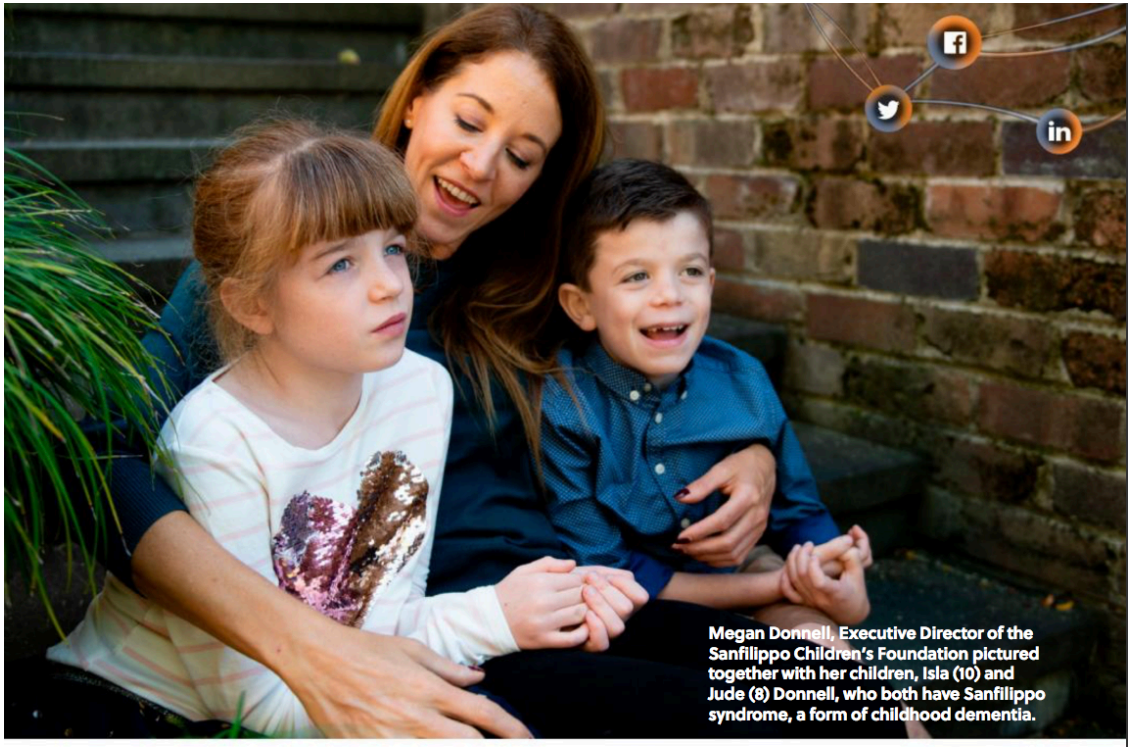
The Sanfilippo Children's Foundation will fund the $2.5 million project having worked with the researchers to secure $2 million from the Federal Government's Medical Research Future Fund (MRFF) and chipping in a further $500,000 of its own funds.

Dr. Nicholas Smith, the head of the Paediatric Neurodegenerative Diseases Research Group University of Adelaide and the Paediatric Neurology Service at the Women's and Children's Hospital in Adelaide, is the project's other Chief Investigator. He welcomes the funding as a demonstration of the Government's commitment to invest in rare disease research.

A multi-disciplinary team will lead the project. Pictured (L to R) Rear - Dr Nicholas Smith, Megan Donnell, Professor Mark Hutchinson, and Dr. Cedric Bardy.



▲ *Reverse engineering patient skin cells into neural (brain) cells to be used as a tool for therapy development*



Megan Donnell, Executive Director of the Sanfilippo Children's Foundation pictured together with her children, Isla (10) and Jude (8) Donnell, who both have Sanfilippo syndrome, a form of childhood dementia.

"Sufferers of rare diseases are historically underserved from a research perspective, particularly in paediatrics," Dr Smith said.

"This vital work can not only improve the lives of those suffering with Sanfilippo but has the potential to yield findings with far-reaching clinical influence on many more common neurological diseases."

STATE OF THE ART

The two-year research project will also involve neurobiologist Dr Cedric Bardy from SAHMRI and Flinders University and Professor Mark Hutchinson from the University of Adelaide.

A variety of techniques will be used by the multidisciplinary team to study the health of the patient-derived neural cell models in comparison to those from healthy participants, and how they respond to compounds. This will include state of the art neuronal anatomy imaging, electrophysiology and transcriptome analysis.

Professor Hutchinson brings a highly innovative aspect to the project – BioPhotonics - a rapidly emerging field with a wide range of applications in clinical medicine and biology. It involves analysing the endogenous fluorescence of biological material, and in this instance, it will allow real-time monitoring of structural and metabolic processes on-going within the neural cells.

AN UNMET NEED

Sanfilippo Children's Foundation Executive Director Megan Donnell says her organisation's purpose is to ease the considerable burden on children with Sanfilippo and their families.

"On average, five Australians each year are born with this condition which is currently untreatable, let alone curable."

"We are thrilled to be partnering with the Government and world-leading researchers in Adelaide. We hope this ground-breaking method of personalised drug screening can help improve the quality of life for children battling Sanfilippo.

> ***"This project is a true demonstration of collaboration with four organisations involved. Alone we can do so little; together we can do so much," Ms Donnell said.***

Sanfilippo Syndrome, also known as Mucopolysaccharidosis (MPS) III, is a serious degenerative condition that causes fatal brain damage. The condition presents in early childhood following an initial period of normal development.

Over time, sufferers experience declining brain function with hyperactive behaviour, disordered sleep, seizures, progressive dementia and loss of mobility. Life-expectancy for children with Sanfilippo is between 12 and 20 years.

Author: The Sanfilippo Children's Foundation